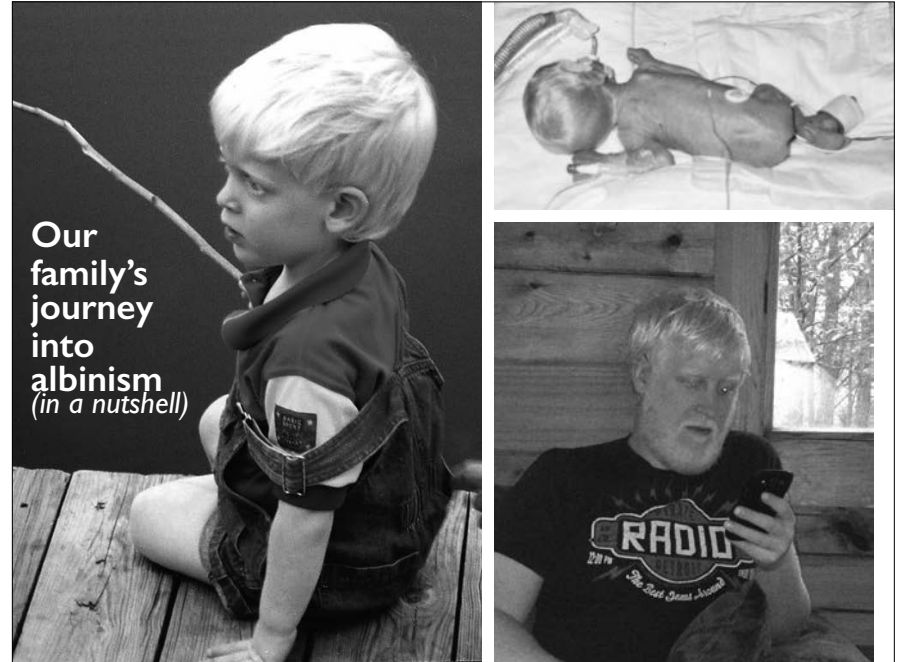


ALBINISM  
in  
HUMANS  
and  
WILDLIFE  
*Myths vs. Truths*

by Lynn Oliver,  
VA wildlife rehabber  
and mom



Our family's  
journey  
into  
albinism  
*(in a nutshell)*

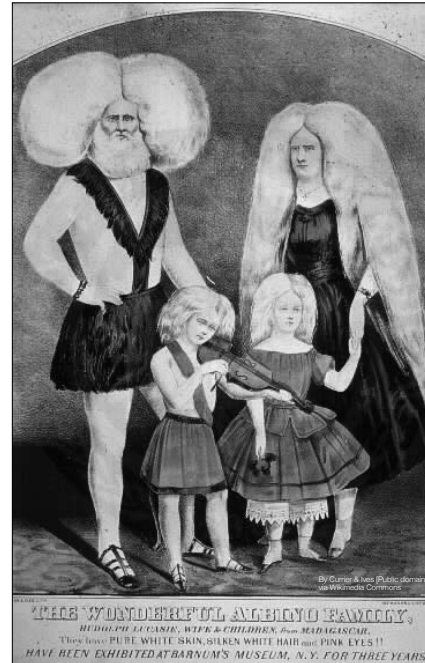


albinism.org

The world of albinism is fraught  
with myth and misinformation



Humans with albinism are frequently vilified in the media. They are presented as evil, or stupid, or otherwise mentally “off.” Or they are displayed as freaks.



The condition shouldn't define the person or animal. First, they are STILL the person or animal they ARE, only with a “condition.” A person with likes and dislikes, goals and dreams; and an animal with a will to live, reproduce and thrive in the wild.

“Albino” is derived from the latin word for white: “Albus”

Something you should know:

In the U.S., humans who have this condition consider it rude to be called “albino.” It's more proper to say “a person with albinism.”

(Although, they reserve the right to call THEMSELVES albinos when joking.)



Albinism occurs in humans AND animals.

Both parents must carry a recessive gene.

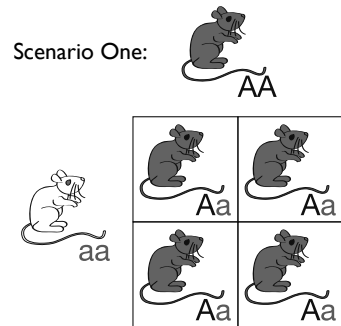


Photo courtesy of Positive Exposure, Rick Gustoff

## Albinism is, at it's basic core:

The result of a gene that suppresses the formation of melanin.

### Basic genetic inheritance patterns

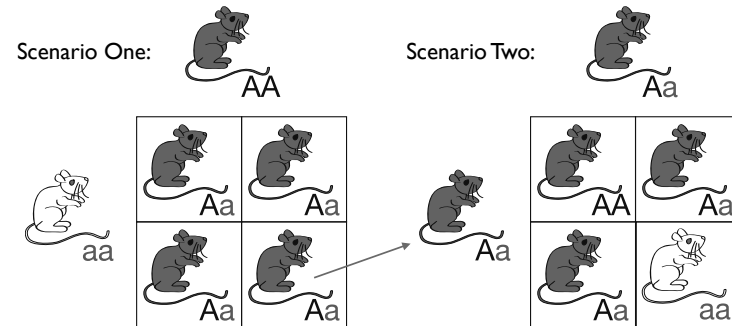


If an animal with two copies of the recessive albino gene mates with an animal with two normal copies, each of it's offspring will have traditional coloring.

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If an animal with two copies of the recessive albino gene mates with an animal with two normal copies, each of it's offspring will have traditional coloring.

If both parents have one copy of the recessive gene for albinism when they mate there is a one-in-four chance that their offspring will display albinism.

## Frequency

- Albinism in humans occurs in approx one in 17,000–20,000 births.
- It occurs more in animals: one in 10,000 for mammals.
- One in 2,000 for birds.
- Cannot find data on frequency in reptiles or fish.



Photos from Suzanne McBride



Photos from <https://www.facebook.com/Albinism>





Photo from <https://www.timesrecordnews.com/photos/galleries/2008/jun/13/albino-raccoon-sighting/>  
enlarged

## Siblings



Photo courtesy of Susan Leslie DuBois



Photo courtesy of Positive Exposure, Rick Guidotti

Photo courtesy of Positive Exposure, Rick Guidotti



Photo courtesy of Amanda Rodriguez

Oculocutaneous = OCA, or Eyes, Skin, Hair

## How albinism affects the skin and hair

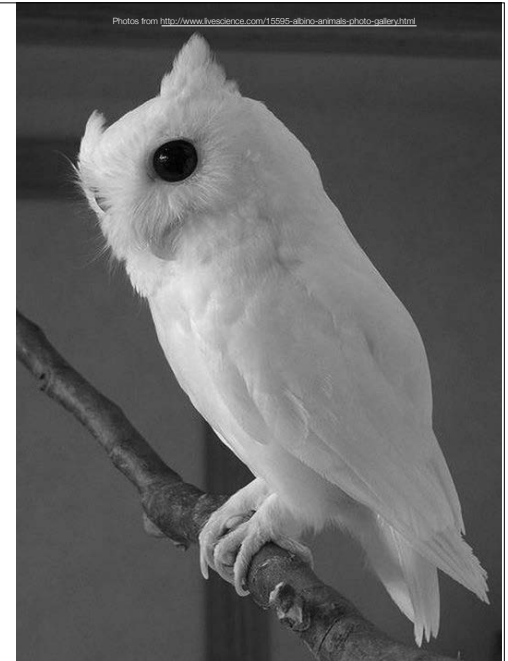
Generally, humans with albinism have flax-white hair, blue eyes and pale white skin which makes them stand out. Sometimes hair pigmentation is not completely absent (white) but shows a pale or medium blonde. Often the affected persons are paler in complexion than the rest of the family. Animals with albinism are similarly pale or shades of yellow/orange/white.





## Not just ONE type of albinism

- “OCA” means Ocular Cutaneous Albinism, or eyes, skin and hair lack pigment.
- OCA1 is caused by genetic mutation of the amino acid tyrosinase which is converted into melanin. OCA1 is further broken down into 1a and 1b. People with OCA1a have no pigment, white hair and light blue, gray or violet eyes. People with OCA1b have detectable levels of pigment.
- OCA2 is caused by a different gene that governs another enzyme for melanin production. People with OCA2 have more pigment and better vision.
- There are also OCA3, OCA4, OCA5, OCA6 and OCA7 which are less common. And, there’s OA (Ocular Albinism), which only affects the eyes and “mostly” occurs in males.
- OCA1, OCA2, OCA4 are seen in the animal world.



Name	Gene	Description
OCA1	TYR	<p>Associated with reduced production of melanin in the skin, hair and eyes. There are two types of OCA1. Individuals affected with OCA1A have a complete absence of melanin pigment resulting in white hair and white skin at birth and irises that do not become darker over time. Visual acuity in individuals can range from 20/200 to 20/400. Individuals with OCA1B have white or light yellow hair at birth that can darken over time, white skin that darkens over time and irises that may change from light blue to green or brown over time. Vision is usually better in individuals with OCA1B than in those with OCA1A.</p> <p>OCA1 is associated with abnormalities (mutations) in the tyrosinase (TYR) gene. The TYR gene is responsible for the production of the enzyme tyrosinase which is the key enzyme in the formation of melanin pigment. Some TYR mutations result in the production of a completely nonfunctioning tyrosinase enzyme and no melanin pigment is formed. This results in OCA1A. Different TYR mutations result in the production of a tyrosinase enzyme with limited enzymatic activity but it is still able to produce small amounts of melanin pigment. This type of OCA1 is called OCA1B. In the case of OCA1B, melanin pigment will accumulate with time in the skin, hair and eyes.</p>
OCA2	OCA2 P Gene	<p>Associated with the same vision problems that occur in OCA1. Individuals with OCA2 have a wide range of skin pigmentation that is partially dependent on their genetic background of the affected individual and the mutations present. Hair color is usually not completely white and there can be some pigment present in the skin but skin color is usually lighter than in unaffected relatives. Individuals with extensive sun exposure can develop pigmented nevi and lentigines (dark spots on the skin). This does not occur with other types of OCA. A reduction in skin pigment is apparent in Africans and African-Americans but skin coloration appears close to normal in other populations with normally lighter skin pigmentation but affected individuals do not tan. Brown OCA is a type of OCA2 where hair and skin coloration is darker. This type of OCA2 has only been reported in individuals with African ancestry.</p> <p>OCA2 is associated with mutations in the OCA2 gene (also called the P gene). The OCA2 gene is responsible for production of the OCA2 protein. The precise function of the OCA2 protein is unknown, but it is thought to be important in regulating the movement of the substrate tyrosine into the melanosome as well as regulating the internal environment of the melanosome.</p>
OCA3	TYRP1	<p>Initially described in the African population. Affected individuals have red to reddish-brown skin, ginger or reddish hair, and hazel or brown eyes and the condition was initially termed rufous albinism. OCA3 has now been identified in several additional populations including those of Asian descent (Chinese and Japanese), Asian Indian and Northern European. Affected individuals of Asian heritage can have blond hair with light brown eyebrows with skin lighter than their parents. Both hair and skin pigmentation increases with age. Reduction in visual acuity is not as severe as in OCA1 or OCA2. Nystagmus and photophobia may not be present.</p> <p>OCA3 is associated with mutations in the tyrosinase related protein 1 (TYRP1) gene. This gene is responsible for the production of tyrosinase-related protein-1, an enzyme like tyrosinase, which is involved in the production of melanin. The TYRP1 enzyme is part of a gene family that includes tyrosinase and the tyrosinase related protein-2 (TYRP2), all of which are enzymes involved in melanin biosynthesis. The TYRP1 enzyme is responsible for later steps (after the initial tyrosinase step) in melanin pigment production.</p>

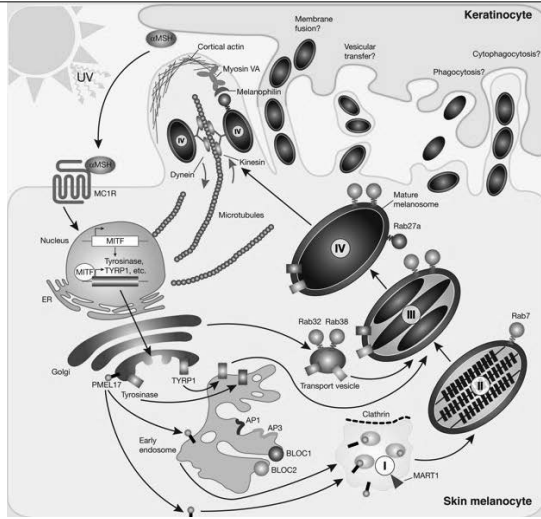
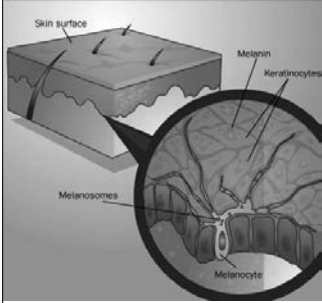
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[http://realhistorywww.com/world\\_history/ancient/Misc/Data/Comprehensive\\_data\\_on\\_albinism.htm](http://realhistorywww.com/world_history/ancient/Misc/Data/Comprehensive_data_on_albinism.htm) (warning: a little big of a nutcase!)

Name	Gene	Description
OCA4	SLC45A2 (membrane-associated transporter protein; MATP)	<p>Characterized by physical features that are similar to those of OCA2. Hair color of affected individuals can range from yellow to brown. Visual acuity can range from 20/30 to 20/400 depending on the amount of pigment that is present, but acuity is usually in the range of 20/100 to 20/200. OCA4 was initially identified in an individual of Turkish origin and has been also found in Asian populations including Japanese and Korean and German individuals.</p> <p>OCA4 is associated with mutations in the SLC45A2 gene (also called the membrane-associated transporter protein; MATP). The SLC45A2 gene is responsible for the production of a membrane associated transporter protein formed with 12 transmembrane helices. The precise function of this protein is unknown but it is required for the normal production of melanin by the melanocyte.</p>
OCA5	4q24	<p>Has been found in only one family in Pakistan. Affected individuals have golden colored hair, white skin and the same visual problems that occur in OCA1. Visual acuity in this family was 6/60.</p> <p>The gene responsible for OCA5 has been located on chromosome 4 (4q24). 14 genes are in this location, but the specific causative gene for OCA5 has not yet been determined.</p>
OCA6	SLC24A5	<p>Characterized as having golden to light to dark brown hair, white skin and brownish irides and has been classified as autosomal recessive ocular albinism (AROA), though individuals are hypopigmented when compared to their parents. Only a few individuals have been identified with this type of albinism and all of the clinical features of OCA6 have not been determined but it is assumed that the reduction in visual acuity will not be as severe as seen in OCA1.</p> <p>OCA6 is associated with mutations in the SLC24A5 gene. The SLC24A5 gene is responsible for the production of a membrane associated transporter protein. The precise function of this protein is unknown but it belongs to a family of potassium-dependent sodium/calcium exchangers. It may be involved in the maturation of melanosomes.</p>
OCA7	C10orf11	<p>Characterized with blond to dark brown hair and skin which is more hypopigmented than parents. Individuals had nystagmus and iris transillumination. Visual acuity ranges from 6/18 to 3/60.</p> <p>OCA7 is associated with mutations in C10orf11. The isoform 1 open reading frame encodes a 226 amino acid protein containing a leucine-rich repeat. The function of the protein is unknown but is thought to play a role in melanocyte differentiation.</p>

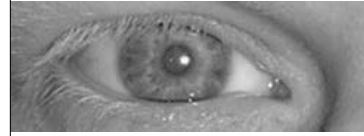
<https://rarediseases.org/rare-diseases/oculocutaneous-albinism/>  
[http://realhistorywww.com/world\\_history/ancient/Misc/Data/Comprehensive\\_data\\_on\\_albinism.htm](http://realhistorywww.com/world_history/ancient/Misc/Data/Comprehensive_data_on_albinism.htm) (warning: a little big of a nutcase!)

# Melanin and it's relationship to Albinism

## How Albinism Works



# How albinism affects the eyes.



By Karen Gornikow, Jakob Ek, and Karen Brondum-Nielsen [CC-BY-2.0] [www.researchgate.net/publication/228424246](http://www.researchgate.net/publication/228424246), via Wikimedia Commons

## Myth: albinos have "red" eyes.

- The truth is, people and animals with albinism can have blue, gray, hazel or other lighter colored eyes.
- We see "red" eyes because we see the blood-rich vessels through the irises and into the retinal wall due to lack of pigment.



# In humans, vision issues are widely varied from "legally blind" to "able to drive."

- All have some visual issues, but 5% of people with albinism have enough pigment to see well enough to drive without correction.



Photos courtesy of Positive Exposure, Rick Guidotti



Photo courtesy of Bat World Sanctuary



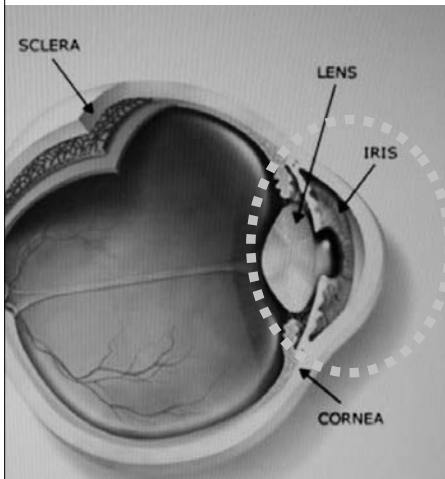
Photo from Brenda Burwell, DVM



Photo from <http://www.liveasapet.com/15695-albino-squirrels-photo-gallery.html>

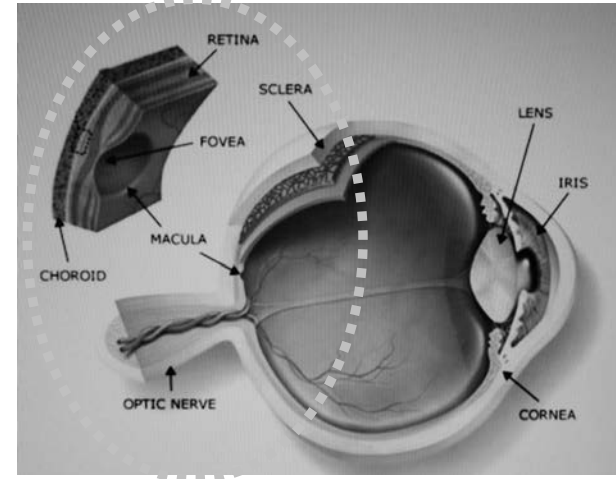
## Photophobia

- Reduced pigment in the iris causes photophobia (sensitivity to sunlight).



## The retina

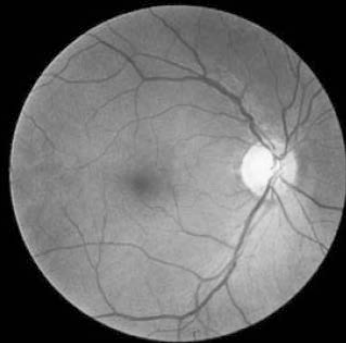
The primary reason people with albinism can't see as well as normally-sighted people is because they have fewer cones in the fovea which is within the macula. In other words, they have fewer "dots" to make up the picture we see (think HDTV vs. regular TV). The cones in particular are the "dots" that people use to see details, such as printed letters and numbers or details of people's faces.



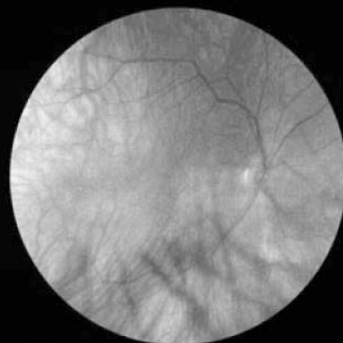
## The retina

From presentation "How We See - Albino Eye" by Brian Evans

### Pigmento

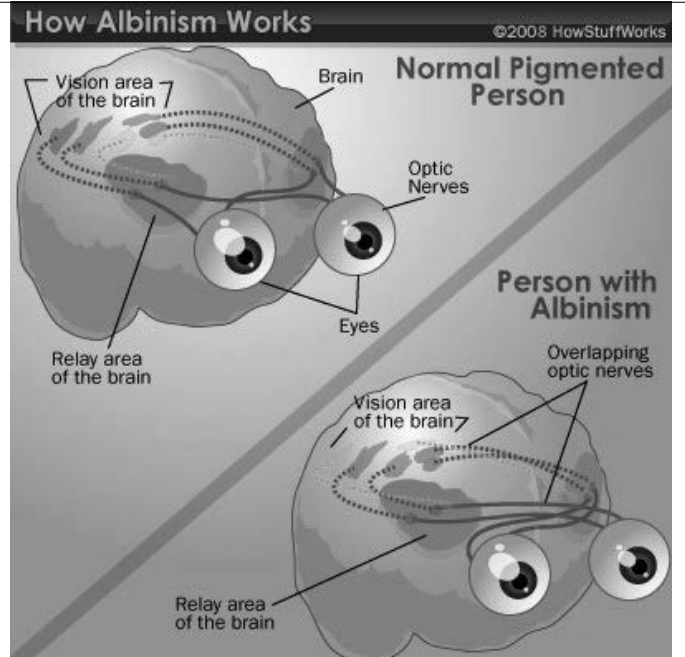


### Albino



## The optic nerves

- The optic nerves rewire differently in utero (directed by melanin) from normally pigmented people and can cause strabismus (eyes not lined up, resulting in crossing or outward turning).

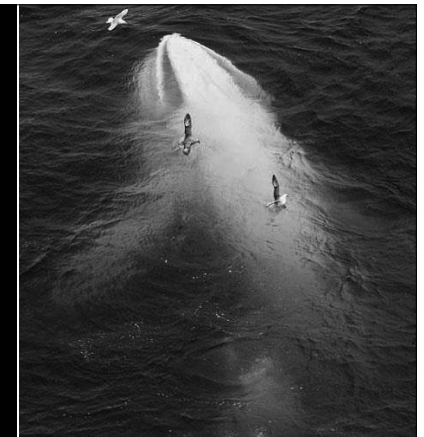






## Plants can have albinism, too!

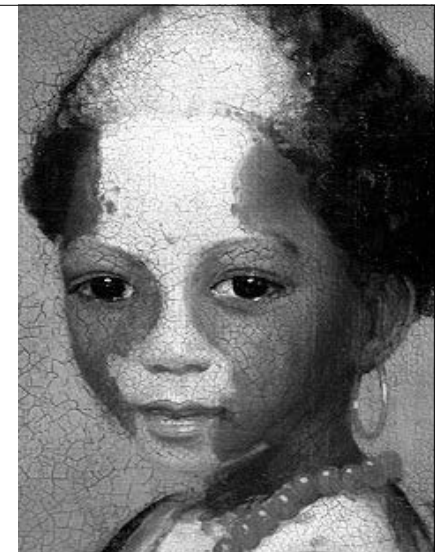
- **Missing pigment called chlorophyll:** They generally don't live long because without chlorophyll, they can't convert the sun's energy into growth.



## The wide world of skin/eye/hair/fur/feather color in the human/animal kingdom

### Conditions similar to albinism

- **Piebaldism:** animal is one that has a spotting pattern of large unpigmented, usually white, areas of hair, feathers, or scales and normally pigmented patches, generally brown or black.



- **Vitiligo/Hypopigmentation:** A condition in which the skin is partly brown and partly white.

- **Leucism:** is much like albinism, but it is caused by a reduction in all types of pigmentation, not just melanin. Also, because eye cells are derived from an independent developmental origin that isn't affected by leucistic phenotypes, leucism does not affect eye color. Leucism though, may not affect the whole body, leaving certain patches as normally pigmented.

**Other interesting pigment anomalies:**

- In **erythristic** animals, red replaces other forms of melanin.
- **Xanthochromism** (also called xanthochroism or xanthism) is a term that may be applied to animals whose coloration is unusually yellow through an excess of yellow pigment, or possibly a loss of darker pigments that allows yellow pigment to be unusually dominant.
- Some birds obtain their red or yellow color from carotenoids in their diet. The flamingo is a good example of this.



**Conditions “opposite” to albinism:**

- **Melanism:** the occurrence of an increased amount of dark pigmentation of skin, feathers, eyes or hair in an organism, resulting from the presence of more than normal melanin. It is the opposite of leucism and albinism.



**What about a calico cat?**

Coat color in cats is a sex-linked trait, a physical characteristic (coat color) related to gender. Female animals have two X chromosomes (XX), males have one X chromosome and one Y chromosome (XY). The genetic coding for displaying black or orange color is found on the X chromosome. The coding for white is a completely separate gene.

Since females have two X chromosomes, they are able to “display” two colors (orange and black, or variations thereof) and white; creating the 3-color calico mix. Since males have only one X chromosome, they can only be orange OR black. It is more complicated than simply having the color genes -- it is a complex process of dominant and non-dominant genes interacting on the X chromosomes, but that is the basis for coat color in calico cats.

Can a calico cat ever be male? Yes, in rare instances. In this situation, the cat has two X chromosomes and one Y chromosome (XXY). Cats with this chromosomal configuration are usually sterile (not able to breed).



**What are the advantages and disadvantages of having albinism?**

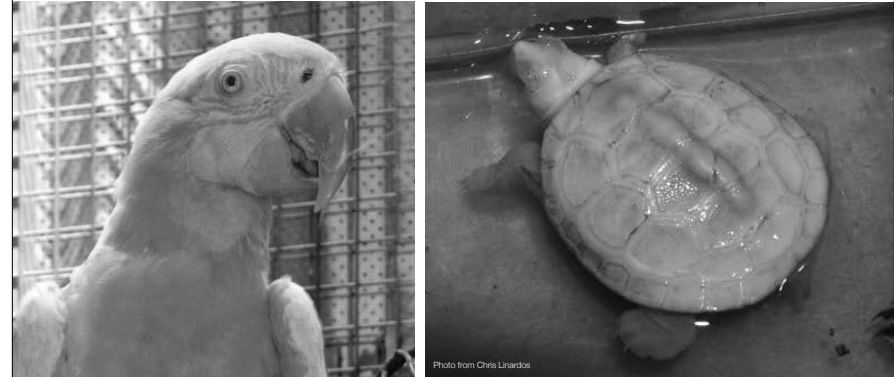


### Disadvantages of having albinism

- Easy prey; easily seen and tracked by predators (or if a predator, easily seen and avoided by prey).
- Many albino animals face a higher risk of skin cancers and retinal damage without melanin to protect them. In the case of some albino reptiles, species that bask in the sun to warm themselves, sunlight may quickly prove fatal because they don't realize they are burning.
- Animals with albinism can have visual issues similar to humans: nystagmus and strabismus and low vision.
- Sometimes albinism is caused by other disorders, like in humans, Chediak-Higashi, which have immune damaging effects.
- Albinism also may make life more difficult for some birds and other animals that use color to attract mates (but remember, not all animals see the color spectrum that humans see). Several songbird females select males based on their courtship displays. Having a display missing a crucial splash of color may put the animal at a competitive disadvantage.



### When you have albinism, you burn, not tan.



**RIGHT NOW IN AFRICA:** People with albinism are being hunted, maimed, dismembered and killed because witch doctors are spreading the myth that having a body part from an albino person will bring fortune and good luck. Please read more on this at Asante-Mariamou.org and at <http://www.albinism.org>.



These ideas about people with albinism have no scientific basis and are untrue:

They are lucky, magical, or can cure diseases.

They are evil or stupid.

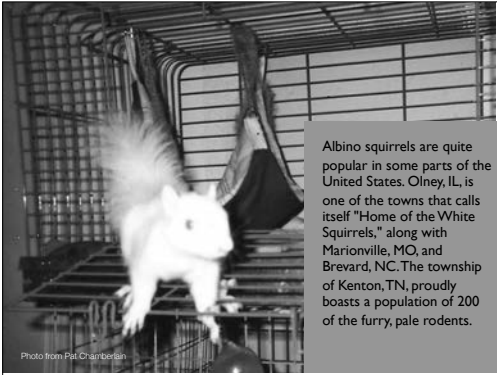
They are the result of incest. (They are the result of mating between two people who carry a mutation in the same melanin-related gene. Most often, the parents are unrelated.)



### Benefits of having albinism

- Because they are rare, albino animals have often been given mythical status. Many Native Americans, for example, considered white buffalo to be sources of immense power and good fortune. To do harm to them would bring misfortune. In Australia, a white (hypo-pigmented) humpback whale named Migaloo (native for "white fellow") is honored by Aborigines.
- Animals that are legal to be bought or sold can bring a higher price if they are albinos. Breeders of amphibians and reptiles for captive animal markets often test and select for albino offspring. Several zoos proudly keep albino specimens.
- Because they lack color, albino animals have a ghostly beauty. Many people count themselves lucky to see one. You can increase your chance of discovering one of these rare oddities of nature by spending more time outdoors.
- Makes it easy to find your kid in a crowd (unless it's a conference with 800 people with albinism!)
- Lack of pigmentation leads to abnormal development of their eyes and poor vision for details. The good news? "*Animals in the wild rely on peripheral vision to spot movement. And that remains intact,*" says Dr. Rick Thompson, an eye doctor and chair of the board of scientific advisers for the National Organization for Albinism and Hypopigmentation (NOAH).





Albino squirrels are quite popular in some parts of the United States. Olney, IL, is one of the towns that call itself "Home of the White Squirrels," along with Marionville, MO, and Brevard, NC. The township of Kenton, TN, proudly boasts a population of 200 of the furry, pale rodents.



Photo from Jeanette Wright, courtesy of Norman Wiley and Jerry Bondell

• In the South Pacific island nation of Fiji, people with albinism are revered and have historically held high-ranking positions within the community.



Photos from Chris Linardos

Now that you know a bit more about what albinism is...



## Rehabber's dilemma: to release or not?

Based on what we know, vision acuity is low, therefore it makes sense that not only are albino wildlife easier to BE seen by predators, but they are less likely to SEE predators. BUT their peripheral vision remains intact, so their ability to spot movement can save them.

On the other hand, they often compensate with other senses like hearing. Obviously, enough survive to pass on the genes, so I would venture to say it needs to be handled on a case-by-case basis, dependent upon your observations of how well the animal has achieved all the other release criteria.

## What do YOU think?



## Resources:

- National Organization for Albinism and Hypopigmentation (NOAH) with information from their two books on children with albinism <http://www.albinism.org>
- Positive Exposure <http://www.positiveexposure.org>
- Asante Mariamu <http://www.asante-mariamamu.org>
- Bonnie S. LeRoy, MS, CGC  
Editor, Journal of Genetic Counseling  
Director of the Graduate Program of Study in Genetic Counseling  
Associate Professor; Genetics, Cell Biology and Development  
University of Minnesota
- Bill Oetting, a researcher at the University of Minnesota (Bonnie LeRoy's colleague, who helped answer questions)
- Understanding more about pigmentation: <http://ics.biologists.org/content/121/24/3995.full>
- <http://www.thetexasherp.com/2011/08/quick-facts-weve-most-all-have-heard-of.html>
- [http://findarticles.com/p/articles/mi\\_m1590/is\\_6\\_61/ai\\_n9478507](http://findarticles.com/p/articles/mi_m1590/is_6_61/ai_n9478507)
- [http://wiki.answers.com/Q/Who\\_does\\_Albinism\\_commonly\\_affect](http://wiki.answers.com/Q/Who_does_Albinism_commonly_affect)  
(Plus all the photo links in small print)
- NORD Rare Disease Database: <https://rarediseases.org/rare-diseases/oculocutaneous-albinism/>
- <https://onlinelibrary.wiley.com/doi/abs/10.1111/j.1600-0749.1998.tb00713.x>
- <https://www.ncbi.nlm.nih.gov/pubmed/865082>



## My info:

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540-465-5315 – [lynn@valleywildlife.org](mailto:lynn@valleywildlife.org)
- I live near Strasburg, VA
- Part of a nonprofit organization with several other rehabbers called Valley Wildlife Care
- Mostly rehab small mammals: squirrels, flyers, chipmunks and groundhogs and skunks (generally BABIES-only for skunks).